

COMPLEX LEARNING DIFFICULTIES AND DISABILITIES RESEARCH PROJECT (CLDD)

FRAGILE X

What is Fragile X Syndrome?

Fragile X syndrome is the most common inherited form of learning disability. It is a chromosomal disorder caused by a 'fragile' site on the end of the X chromosome – appearing to be breaking, but is not quite separated. The gene which causes fragile X syndrome has been identified as the FMR1 gene. It is a gene present in everybody, but an increase in the size of part of the gene, or a mutation, can prevent it from working properly, thus causing learning disability. Those affected with fragile X syndrome have a full mutation. Those with a small change, (a permutation), of FMR1 gene are carriers of fragile X syndrome, but are not necessarily affected by it. Both men and women can be carriers of a premutation gene, and the syndrome can occur in both sexes in all populations. However, it is more prevalent in males than females.

Possible indicators of Fragile X Syndrome

Distinctive facial features may include a large head with long face, large jaw, prominent ears, a long and flattened nasal bridge, and a high arched palate often with dental overcrowding. Other indicators are connective tissue problems such as flat feet, double jointedness, soft skin and spine curvature, which can become more pronounced with age. However, despite all of these potential characteristics, children often do not have an unusual appearance and the characteristics may not be present.

Fragile X syndrome can cause learning difficulties from mild to severe. It can also cause a range of additional difficulties including:

- social communication difficulties
- speech and language difficulties
- attention and emotional difficulties
- behavioural problems
- floppy muscle tone or hypotonia
- developmental delays
- sensory integration difficulties
- seizures
- at the onset of puberty (often premature), enlarged testicles in males and menstrual problems in females; the potential physical and social discomfort of this should be considered, especially within the everyday pressures of school.

Some girls with fragile X syndrome may be affected by learning difficulties to a lesser extent than boys. Many typical profiles of students with fragile X syndrome are similar to those of students with Autistic Spectrum Disorder (ASD), and it is estimated that 25–35% of young children with fragile X syndrome have an additional diagnosis of ASD; 70–90% of boys and 30–50% of girls with fragile X syndrome have attention deficit hyperactivity disorder (ADHD). For most, there will just be a cross-over in characteristics.



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Implications for teaching and learning

These students may have **strengths** in:

- expressive language, receptive vocabularies including verbal labelling
- short and long-term memory for meaningful information, including good visual memory for environment around them
- recognising and understanding emotional expression in others
- being friendly, helpful and curious, with a good sense of humour (particularly boys).

Difficulties for students may present as follows:

- working memory difficulties when considering abstract concepts
- organisational /sequencing issues
- eye and vision problems, such as strabismus or 'wobbly eye'
- squints and poor focus on work may occur if sensitivity to touch prevents students from wearing glasses comfortably
- ear infections or 'glue ear'
- repetitive use of language; discussion revolving around favourite topics regardless of appropriateness
- hyperarousal and anxiety when faced with the social demands of language, such as eye contact, coordination of syntax, semantics and conversational pragmatics, leading to an inability to plan
- verbal responses
- outbursts, tantrums and even aggression in boys; girls tend to be less hyperactive and have more mood stability than boys
- gross and fine motor difficulties mean that students have difficulty with handwriting, dressing, manipulating tools and eating (compounded by poor oral-motor co-ordination)
- difficulties integrating any two or more sensory inputs at one time can lead to behavioural problems due to overload
- tactile defensiveness is very common in these students; harmless sensations are perceived as potentially dangerous and therefore unpleasant; sensitivity to being touched lightly may be perceived as painful
- hypersensitivity may mean that students dislike art activities such as painting or using clay, or carrying out daily care or hygiene tasks involving washing hands or shampooing hair
- difficulty filtering out peripheral noises, for example, outside traffic or others talking quietly affects concentration; if there is too much going on, the situation can seem confusing, resulting in anxiety and possibly challenging behaviour
- oral sensitivity can result in food with an unusual texture or taste, or the use of particular cutlery, being unpleasant for these students, which can affect behaviour at mealtimes
- hypersensitivity to smells can provoke heightened positive or negative responses ranging from extreme preoccupation to aversion
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- speech may be hard to understand, particularly when speaking for longer periods of time; it is often delivered in short bursts followed by long pauses, with repetitions of words or phrases.

Girls with fragile X syndrome are likely to show particular difficulties related to organising their thoughts, planning ahead and shifting between topics; extreme shyness and anxiety in social situations; oversensitivity to perceived criticism and rejection; difficulty in picking up signals in social situations; difficulty in seeing the consequences of their actions, which impacts on their self-esteem, and their ability to make friends, however much they want to.

Boys can experience delay in language acquisition, excessive repetition of words or phrases, impulsive speech and poor pragmatic skills; poor short and long-term memory for abstract and non-meaningful information (related to poor function of working memory); attention and concentration problems; and arithmetic, in particular processing and recalling sequential and abstract information. Whilst they may be good in a conversation in terms of their knowledge and vocabulary, their sequencing of ideas might get muddled, and they may struggle with appropriate turn-taking.

Supporting students with fragile X

The following approaches may be effective:

1. For abstract information, give small chunks of information at a time, focusing on students' strengths in verbal skills and good memory in a meaningful context.
2. Breaking things down into small steps may not always be best practice for these students as they may need to see a more complete picture of what they are learning. For example, when learning to read, try building up a sight vocabulary, putting whole words in a meaningful context; phonics require abstract thinking and memory, sequential processing, and sight and sound integration – areas of difficulty for some boys with fragile X syndrome.
3. Visual aids in teaching and more practical-based approaches may make tasks more concrete and easier to engage with, rather than more detailed spoken instructions. ICT based approaches might be beneficial as material can be delivered visually, with an instant response to an action, but also avoiding direct teacher interaction, which some students may find hard to cope with.
4. Tasks need to be made clear, achievable and able to be completed in a reasonable time span. Students will benefit from a focused, structured and predictable routine with change kept to a minimum.
5. Direct instructions may be better given whilst sitting or standing alongside the student, rather than in front of them, to avoid expectation of eye contact.
6. Frequent states of anxiety and/or hyperarousal will require calming strategies, particularly when out in the community or contexts which present challenges to their senses.
7. Often, these students will not only avoid stimuli, but also seek sensory experiences to calm or to heighten levels of arousal. Deep pressure can produce a calming affect for some students with



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sensory integration difficulties. If this is the case, try massage, use of weighted blankets or jackets, wearing backpacks, or engaging in activities such as hiking, rolling, gardening or games involving smell.

8. Consultation with an occupational therapist may provide a full sensory profile, together with an explanation of proprioceptive (body awareness) and vestibular (balance) senses, and recommendations specific to each student.
9. Lots of praise or a reward system will boost self-esteem and encourage students to focus on their positives and strengths rather than focusing on their difficulties

Key references

A full reference list can be found on the Information Sheet relating to this project.

Fragile X Society (2003) *Fragile X: An introduction to educational needs*. Essex: Fragile X Society.

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